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Single Aberrant Umbilical Artery in a Fetus With Severe Caudal Defects: Sirenomelia or Caudal Dysgenesis

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We describe a 1,000-g twin fetus with absent kidneys and ureters, anal atresia and minimal evidence of external genitalia, and hypoplastic lower limbs with absent feet. A postmortem arteriogram showed a large single umbilical artery in direct continuation with the abdominal aorta, a unique anomaly almost always related to sirenomelia. We discuss the possible diagnosis of this case as sirenomelia or caudal dysgenesis, and the controversy as to whether they are two related or separate entities. Am. J. Med. Genet. 69:409–412, 1997. © 1997 Wiley-Liss, Inc.

KEY WORDS: sirenomelia; caudal dysgenesis; single umbilical artery; postmortem arteriogram

INTRODUCTION

"Sirenomelia" refers to a malformation comprising "fused" or single lower limbs, and urogenital, anal, and lower spine anomalies [Twickler et al., 1993; Stocker and Heifetz, 1987]. Classically, sirenomelia is considered the most severe form of caudal dysgenesis, formerly named caudal "regression" [Duhamel, 1961]. Nevertheless, some authors consider sirenomelia and caudal dysgenesis separate entities [Jones, 1988; Currarino and Weinberg, 1991; Twickler et al., 1993]. We report on a case with nonfused but hypoplastic lower limbs, and a single and very large umbilical artery in direct continuation with abdominal aorta, a unique anomaly almost always related to sirenomelia [Stevenson et al., 1986; Stocker and Heifetz, 1987; Twickler et al., 1993].

CLINICAL REPORT

This was the first gestation of a 29-year-old woman, who had previous sterility problems and became pregnant after clomiphene therapy and artificial insemination with her husbands sperm. At 34 weeks of gestation she was referred because of twin pregnancy. An ultrasonographic examination at that time demonstrated a single placenta with two amniotic sacs. One of the amniotic sacs showed severe oligohydramnios. Maternal serum glucose levels were normal, and urine did not contain glucose. At 35 weeks of gestation a cesarean section was performed. The first twin was a malformed fetus in a breech presentation, weighing 1,000 g, who died 6 hr after birth. The second twin was a 2,250-g

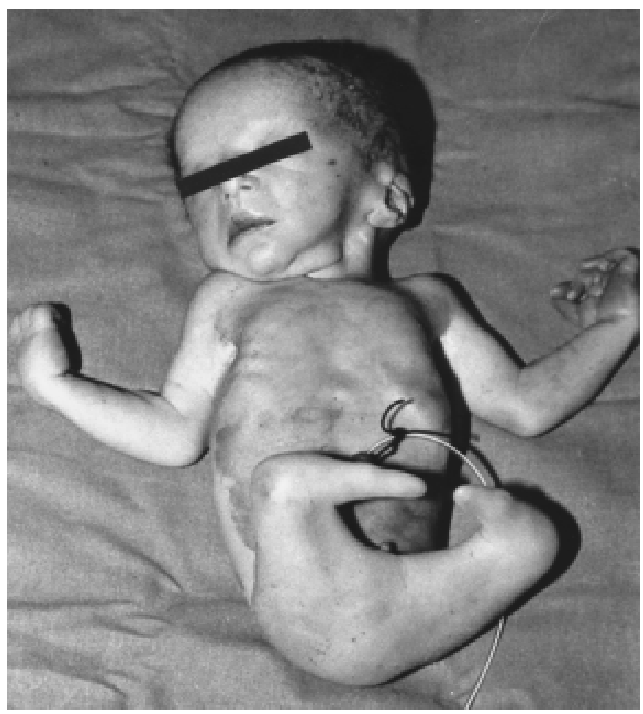

Fig. 1. General (frontal) view of patient. Hypoplastic legs with absence of feet, and anal atresia. Head and upper limbs show typical oligohydramnios deformities but are normally developed.

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Fig. 2. Anterior view of lower part of the body. Legs are separate, with hips in fixed external rotation and knees in fixed flexion. A small skin tag is visible in the genital area. Umbilical cord is catheterized.

phenotypically normal boy, in cephalic presentation, with Apgar scores of 9 and 10 at 1 and 5 min, respectively. His postnatal outcome was uneventful and he was discharged.

The malformed fetus had the typical oligohydramnios (or Potter) sequence with a flat nose, redundant skin folds, overfolded helices of ears, and ulnar deviation of both hands. Lower limbs were separate and severely hypoplastic, with no feet, flexed at knees and externally rotated (Fig. 1), and with contractures at hips and knees. There was anal atresia and no recognizable external genitalia, except for a penis-like skin appendage in the anterior perineal area (Fig. 2). The umbilical cord contained two vessels, both of the same caliber.

A postmortem arteriogram was performed by introducing a catheter through the single umbilical artery. After injection of iodine contrast, an abnormal and very large umbilical artery was seen in direct continuation with the abdominal aorta (Fig. 3). Caudal to this umbilical artery, the aorta continued as a hypoplastic vessel, bifurcating in the pelvic area into two hypoplastic common iliac arteries. Fusion of L3 and L4 vertebrae, hypoplastic sacrum, and a very hypoplastic pelvis were observed (Fig. 4). There was hypoplasia of the tibiae with absence of fibulae.

At necropsy, the lungs were hypoplastic. The diaphragm was complete, and no abnormal findings in liver, spleen, and pancreas were noted. The malformations were all localized to the lower area of the fetus. There were neither kidneys nor ureters. No internal genitalia, bladder, or urethra were identified. The rectum was dilated and attached by a fibrous cord to a cystic mass. This cystic mass was located in the lower abdomen and was full of fluid resembling urine. Microscopic examination showed disorganized renal tissue and intestinal epithelium, both lining the wall of the mass. No gonadal tissue was found in the abdominal cavity.

Pathological examination of the placenta showed an 800-g monochorial, diamniotic placenta with two central umbilical cords. One of the umbilical cords had a single umbilical artery. Specific studies of the vascular connections between placental vessels in both twins were not done.

Chromosome examination from peripheral lymphocytes of the malformed fetus showed a 46,XY normal karyotype.

DISCUSSION

Sirenomelia and caudal dysgenesis are similar malformations [Duncan et al., 1991; Twickler et al., 1993]. There is a long-standing controversy whether both entities are related or if they are different disorders. Some authors consider sirenomelia the extreme manifestation of the caudal dysgenesis "spectrum" [Duhamel, 1961; Davies et al., 1971; Young et al., 1986]. From this point of view we could consider our case a severe form of caudal dysgenesis very close to sirenomelia. However, several authors consider sirenomelia and caudal dysgenesis different entities [Jones, 1988; Currarino and Weinberg, 1991; Twickler et al., 1993]. According to Twickler et al. [1993], caudal dysgenesis is defined by the presence of nonlethal renal anomalies, sacral agenesis, two umbilical arteries, and nonfused lower limbs, while sirenomelia is characterized by the presence of renal agenesis/dysplasia, single aberrant umbilical artery, and fused or single lower limbs. Also, a history of maternal diabetes may be an important

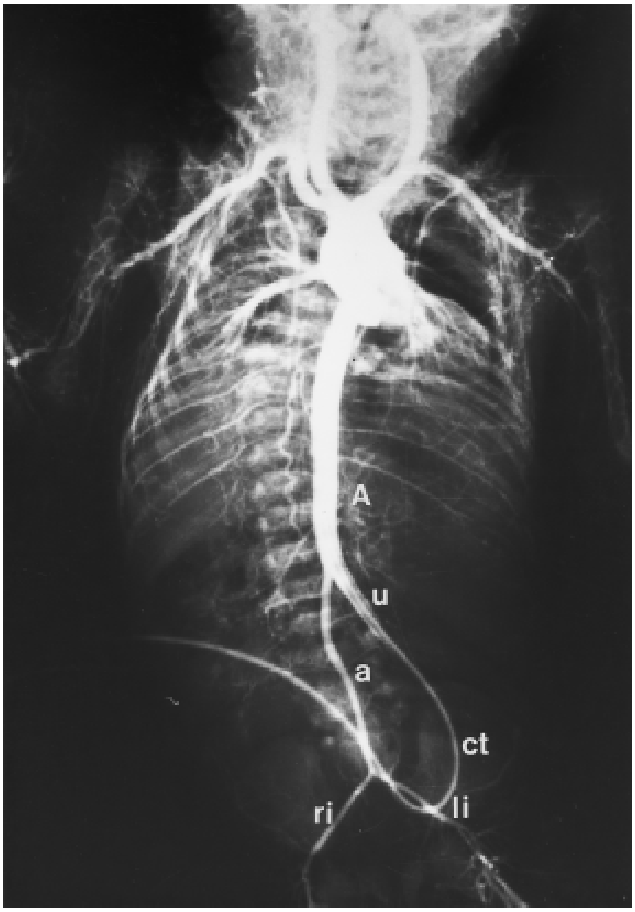

Fig. 3. Postmortem arteriogram through a catheter (ct) inserted in the single umbilical artery. Abdominal aorta (A) connects directly with a large single umbilical artery (u). After the origin of the umbilical artery, the aorta continues as a very thin vessel (a) which then divides into two hypoplastic iliac arteries (ri, right iliac artery; li, left iliac artery).

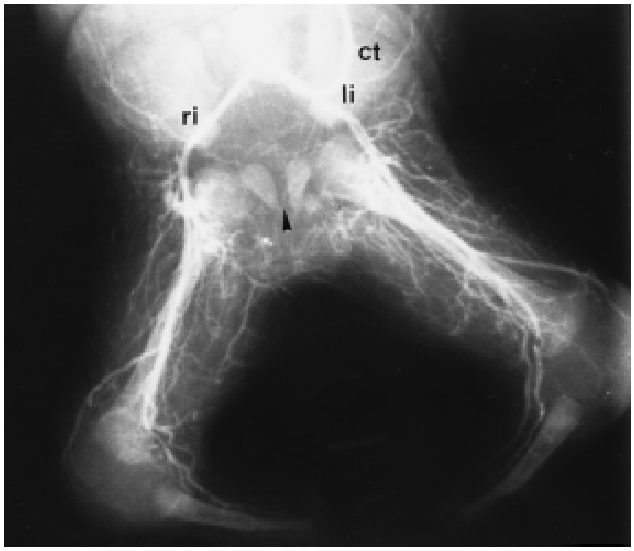

Fig. 4. Arteriogram of lower limbs. ct, catheter; ri, right iliac artery; li, left iliac artery. Arrow indicates contracted pelvis, with ischial bones hypoplastic and very close together. There are two apparently normal femora, with hypoplastic tibiae and absent fibulae.

criterion because it is seldom seen in sirenomelia, whereas it is often associated with caudal dysgenesis [Twickler et al., 1993]. According to this criterion, our case would fit well into the sirenomelia category except for the lack of fused or single lower limbs.

Another finding that favors sirenomelia in our case is the presence of twinning. Monozygotic twining is found in about 15% of cases of sirenomelia [Stocker and Heifetz, 1987]. In our case, both twins were male and the placenta was monochorial and diamniotic, but zygosity studies were not done. The mother became pregnant after clomiphene therapy, and an increased rate of monozygotic twinning has been reported after ovulation induction [Derom et al., 1987]. Thus, monozygosity is highly likely in this case.

Duhamel [1961] mentioned a special case of syrenids, the so-called "anchipod type," with the lower limbs flexed and in external rotation but remaining independent. According to Duhamel [1961], this anomaly was the consequence of an insult specifically localized to the axial portion of the caudal region of the embryo. The case presented here shows lower-limb deficiency with similar characteristics to the Duhamel [1961] anchipod anomaly. In a study of 5 cases with caudal anomalies, Currarino and Weinberg [1991] described an anchipodia case (their case 4). The lower limbs were in fixed flexion at the knees, with external rotation at the hips and with a soft-tissue membrane joining the limbs. This case also presented with a single umbilical artery, but the size and origin of this vessel were not recorded. The rest of the anomalies were almost identical to those of our case, including the presence of a small skin appendage in the area of the external genitalia. Currarino and Weinberg [1991] concluded that caudal dysgenesis constitutes a different entity from a malformation which they designated small pelvic outlet syndrome/sirenomelia (SPOS/sirenomelia). Caudal dysgenesis is characterized by the absence of the sacrum, which in SPOS/sirenomelia is usually less affected or even normal. Imperforate anus, a very constant finding in SPOS/sirenomelia, is a less common anomaly in caudal dysgenesis, even in its most severe forms [Currarino and Weinberg, 1991]. The case presented here fits SPOS/sirenomelia better than caudal dysgenesis because of the imperforate anus, hypoplastic but nonabsent sacrum, and roentgenograms showing a contracted pelvis with small ischial bones very close together.

An important finding in our case is the presence of a single umbilical artery arising directly from the abdominal aorta and then continuing as a large artery with the same caliber as the aorta. Talamo et al. [1982] demonstrated a similar vascular pattern in a sirenomelic stillbirth using the same method, a postmortem arteriogram made through the umbilical artery. This type of vascular anomaly is considered a remnant of the vitelline artery complex and appears as a unique anomaly, almost always associated with sirenomelia [Stevenson et al., 1986; Stocker and Heifetz, 1987; Twickler et al., 1993]. It has also been associated with other malformative patterns with severe caudal anomalies such as limb body wall complex [Palacios

and Rodríguez, 1990], but it has not been demonstrated in caudal dysgenesis.

We consider our case nearer to the "anchipod type" of syrenids [Duhamel, 1961] and the SPOS/sirenomelia [Currarino and Weinberg, 1991] than to caudal dysgenesis. The study of the abdominal vascular pattern in cases of caudal dysgenesis will help to elucidate the nature of these entities. The anomalies in this case evidently arose early during blastogenesis, and maybe considered a single polytopic developmental field anomaly.

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